UBA1 gene

ubiquitin like modifier activating enzyme 1

Normal Function

The *UBA1* gene (also called *UBE1*) provides instructions for making the ubiquitin-activating enzyme E1. This enzyme is involved in a process that targets proteins to be broken down (degraded) within cells. Protein degradation is a normal process that removes damaged or unnecessary proteins and helps maintain the normal functions of cells.

Ubiquitin-activating enzyme E1 is part of the ubiquitin-proteasome system, which is the cell machinery that breaks down unneeded proteins. The ubiquitin-proteasome system acts as the cell's quality control system by disposing of damaged, misshapen, and excess proteins. Ubiquitin-activating enzyme E1 is responsible for the first step in the ubiquitin-proteasome system; it turns on (activates) a small protein called ubiquitin. With the assistance of other proteins, the active ubiquitin attaches to a targeted protein. When a chain of ubiquitin proteins is attached to the targeted protein, the protein is recognized and destroyed by a complex of enzymes called a proteasome.

Health Conditions Related to Genetic Changes

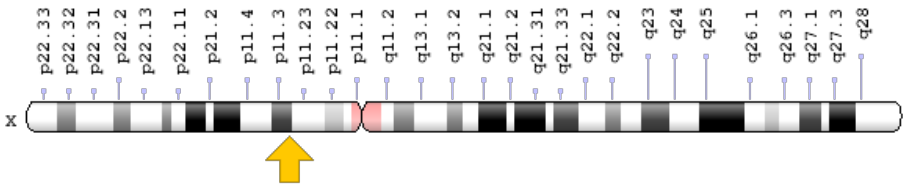
spinal muscular atrophy

At least three mutations in the *UBA1* gene have been found to cause X-linked infantile spinal muscular atrophy. Each of these mutations changes one DNA building block (nucleotide) in the *UBA1* gene. Two of the mutations (written as 1617G>T and 1639A>G) are thought to lead to an enzyme with impaired function. A third mutation (written as 1731C>T) results in a decrease in the activity of the *UBA1* gene, causing less of the enzyme to be produced. Reduced levels of functional enzyme can disrupt the process of protein degradation. A buildup of proteins in cells can cause the cell to die; the motor nerve cells that control muscle movement are particularly susceptible to damage from protein buildup.

Chromosomal Location

Cytogenetic Location: Xp11.3, which is the short (p) arm of the X chromosome at position 11.3

Molecular Location: base pairs 47,190,800 to 47,215,128 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- A1S9
- A1S9T
- A1ST
- AMCX1
- CFAP124
- GXP1
- MGC4781
- SMAX2
- UBA1, ubiquitin-activating enzyme E1 homolog A
- UBA1_HUMAN
- UBA1A
- UBE1
- UBE1X
- ubiquitin-activating enzyme E1
- ubiquitin-like modifier activating enzyme 1

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Ubiquitination/De-Ubiquitination
  https://www.ncbi.nlm.nih.gov/books/NBK6166/#A65133

GeneReviews

- Spinal Muscular Atrophy, X-Linked Infantile
  https://www.ncbi.nlm.nih.gov/books/NBK2594

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28UBA1%5BTIAB%5D%29+OR+%28%28UBA1A%5BTIAB%5D%29+OR+%28UBE1%5BTIAB%5D%29+OR+%28ubiquitin-activating+enzyme+E1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- UBIQUITIN-LIKE MODIFIER-ACTIVATING ENZYME 1
  http://omim.org/entry/314370

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_UBA1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=UBA1%5Bgene%5D
- HGNC Gene Family: Ubiquitin like modifier activating enzymes
  http://www.genenames.org/cgi-bin/genefamilies/set/100
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12469
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7317
- UniProt
  http://www.uniprot.org/uniprot/P22314

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